

The California

Expanded AFP Screening Program

Every pregnant woman wonders about the health of her fetus (unborn baby) and the possibility of birth defects. The Expanded AFP blood test can help detect some birth defects. This booklet describes the test **for women under 35 years of age at delivery**. It is a woman's own decision whether to have the test or not. A consent/refusal form is at the end of this booklet.

The Expanded AFP Screening Program consists of: the Expanded AFP blood test first, followed by diagnostic tests if needed. The Program helps detect open neural tube defects, abdominal wall defects, Down syndrome, and trisomy 18.

Who should consider having the Expanded AFP blood test?

All pregnant women.

Some women need genetic counseling before deciding about this test. If a woman (or the baby's father) has a medical or family history of inherited conditions, she should discuss the test with her doctor. A woman with a high risk pregnancy should also talk to her doctor. There may be special tests that should be done in place of, or in addition to, Expanded AFP.

What does the blood screening test involve?

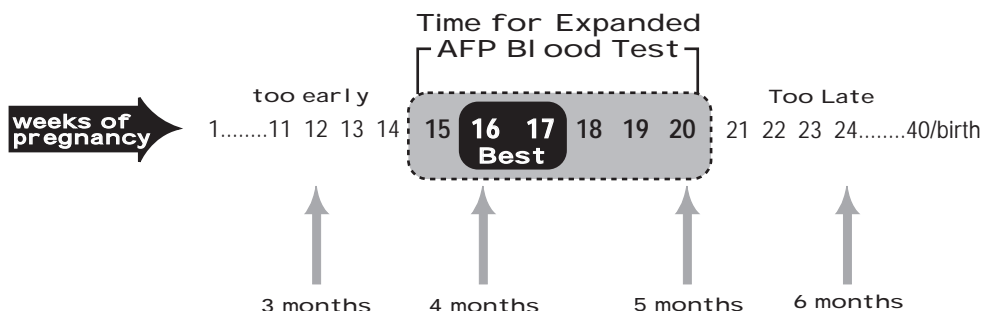
A small amount of blood is taken from the pregnant woman's arm. Her blood is tested for the amount of **AFP** (alpha-fetoprotein), **HCG** (human chorionic gonadotropin), and **UE** (unconjugated estriol). These substances are made by the mother's placenta and the fetus. At each week of pregnancy there are different amounts of these substances in the mother's blood. (What she eats does not affect these substances.)



When is the blood screening test done?

The blood test can only be done reliably **between 15 and 20 weeks of pregnancy**. *The best time is 16 to 17 weeks*. It is important to know how far along the pregnancy is. Ultrasound is very useful for this purpose.

The result of the blood test is sent to the patient's doctor or clinic within 1-2 weeks.



What does a “screen negative” result mean?

It means that the risk for *certain* birth defects is low enough that the Program does not consider follow-up tests necessary. The risk is calculated by measuring the amounts of AFP, HCG and UE in the woman's blood and also by considering her age. About 90% of pregnant women tested will have a “screen negative” result.

Since the blood test is just a screening test, *there is still a chance that the fetus may have a problem* — even when the test result is negative.

What does a “screen positive” result mean?

It means that there is an increased risk for certain birth defects in this pregnancy (such as neural tube defect, abdominal wall defect, Down syndrome, or trisomy 18). The risk is calculated using the amounts of AFP, HCG and UE found in the woman's blood. Her age is part of the calculation for the risk of Down syndrome.

Most of the time, however, the reason for the result is *not* a birth defect. The **most common reasons** for a “screen positive” result include:

- ◆ the due date is earlier or later than thought *or*
- ◆ there is more than one fetus (twins, triplets) *or*
- ◆ the substances in the blood varied more than usual, without any known pregnancy problem.

To determine the reason for the “screen positive” result, **follow-up diagnostic tests are offered** and paid for by the Program. *Most women with “screen positive” results have normal follow-up tests and healthy babies.*

SUMMARY OF TEST RESULTS

“screen negative”	→ No follow-up tests are offered by the Program.
“screen positive”	→ Follow-up tests are provided at no extra cost at a State-approved Prenatal Diagnosis Center.

If the test is “screen positive”, what happens then?

A woman with a “screen positive” result will be **called by her doctor or clinic**. She will be offered diagnostic services at a **State-approved Prenatal Diagnosis Center**. When authorized, these are the follow-up services covered by the Program:

- **Genetic counseling** - A professional counselor discusses the pregnancy and family medical history. Questions are answered to help the woman make decisions about further testing.
- **Ultrasound** - A picture of the fetus is made using sound waves. This picture shows the age of the fetus and whether there are twins. The advanced ultrasound done at a Prenatal Diagnosis Center can also detect certain birth defects.
- **Amniocentesis** - A small amount of fluid is taken out of the uterus by experienced, State-approved doctors. The fluid and the fetal cells in it are tested for specific birth defects.

Women may refuse any of these services at any time.

What if the follow-up tests show that the fetus has a birth defect?

Information will be given to the woman at the Prenatal Diagnosis Center by a doctor or genetic counselor. They will discuss the type of birth defect that has been found and any available treatments. They will also discuss options for continuing or ending the pregnancy. The woman can then make a decision.

The Expanded AFP Screening Program does not pay for any other medical services after the follow-up tests. Referrals for special support services are available.

Birth Defects Found by the Program

What birth defects may be found through follow-up testing?

Open neural tube defects, abdominal wall defects, Down syndrome, trisomy 18, and some other birth defects may be found.

Neural Tube Defects (NTDs)

As a fetus is forming, the neural tube extends from the top of the head to the end of the spine. This develops into the baby's brain and spinal cord. The neural tube is completely formed by 5 weeks after conception.

If there is an opening in the spine, it is called **spina bifida**. This defect often causes paralysis of the legs. It may also cause loss of bowel and bladder control. Frequently, there is water-on-the-brain (hydrocephaly) which requires surgery.

Anencephaly occurs when most of the brain does not develop. This defect causes the death of the fetus or newborn.



Abdominal Wall Defects

Fetuses with these defects have **abnormal openings** on the abdomen. Intestines and other organs form outside the body. Surgery after birth often corrects the defect .

Birth Defects Found by the Program

Down Syndrome

Down syndrome is a common cause of mental retardation. It is associated with heart defects as well. Down syndrome is caused by an extra chromosome #21. Chromosomes are packages of genetic information found in every cell of the body. Birth defects can occur when there are too few or too many chromosomes.

Down syndrome can occur in the fetus of a woman of any age. However, as a woman gets older, her chances increase for carrying a fetus with Down syndrome.

Trisomy 18

Trisomy 18 is caused by an extra chromosome #18. Babies with trisomy 18 have severe mental retardation and usually die before birth or in early infancy.

These birth defects **do not occur very often**. However, **if there is one of these birth defects**, the Expanded AFP Screening Program helps detect it. Among all women who have the Expanded AFP blood test and follow-up tests:

- 97% of the cases of anencephaly are found
- 80% of the cases of open spina bifida are found
- 85% of the cases of abdominal wall defects are found
- 50% or more of the cases of trisomy 18 are found

Among women 35 years of age and under who have the Expanded AFP blood test and follow-up tests:

- 40% to 66% of the cases of Down syndrome are found.

Can the Expanded AFP Screening Program detect every type of birth defect?

No. There are birth defects which **cannot** be detected by Expanded AFP Screening. Even when the blood test is “screen negative”, there is still a chance the fetus may have a problem.

How much does the Expanded AFP Screening Program cost?

The current fee (as of 1995) is **\$115**. This fee may change. (Check with the doctor or clinic about the most current fee.) The fee covers the blood test **and** authorized follow-up services at a State-approved Prenatal Diagnosis Center.

The Program mails a bill to women who have the blood test. Women with private insurance should submit the bill to their insurance company. Women who have a prepaid health plan (HMO) may not receive a bill. If they do, they should send the bill to their health plan office. Women with Medi-Cal usually do not receive a bill. If they do, they should return the bill with their Medi-Cal number or sticker.



If insurance pays only part or none of the bill, the woman is responsible for the bill. Women without health insurance may make monthly payments and are responsible for the whole amount.

Currently, no bill is sent to women with a family history of neural tube defects or to women taking certain seizure medications.

Any charges for drawing blood are not included in the program fee.

If you have questions about the test, ask your doctor.
After you have decided, please sign the consent/refusal
form on the next page.

**Please keep this booklet and your copy of the
consent/refusal form for your records.**



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